

are all highly conserved with respect to the consensus sequence in SLRPs, and these are flanked by cysteine clusters (see Figure 3B) [SEQ ID NO: 2] [10]. The deletion of a portion of the cysteine cluster in the amino-terminal portion of nyctalopin appears to be responsible for complete X-linked CSNB in six families, which highlights the importance of this conserved region. The mutation that causes a stop codon on the carboxyl-terminal side of the leucine-rich repeats and another cysteine cluster, likely affects the ability of the protein to anchor in the membrane, as the protein portion on the carboxyl-terminal side of this mutation is presumed to be important for GPI anchoring nyctalopin in the cellular membrane. Mutations that replace a consensus amino acid with another amino acid are presumed to disrupt an essential amino acid function. Mutations that result in the insertion (or deletion) of amino acids in the protein are presumed to alter the folding of the protein

4. Page 30, lines 6 to 12, change to read:

Six other families were found to have an in-phase 24-nt deletion that results in the loss of eight amino acids - RACPAACA (see Figure 5B). Six of these amino acids form part of a conserved cysteine-cluster on the amino-terminal side of the leucine-rich repeats, as shown in Figure 3B [SEQ ID NO: 2]. Haplotype analysis of X chromosomes with this deletion mutation from each of the six families revealed nearly identical haplotypes, suggesting that these families share a common founder mutation. In three families, insertion mutations representing duplications of adjacent protein sequence add either six or three amino acids (Figure 3B) [SEQ ID NO: 2].

AMENDMENTS TO THE CLAIMS

1. Please amend claims to read as follows

Claims 1-24 (Cancelled)

1 Claim 25 (Previously Added) An isolated or recombinant DNA molecule encoding the amino acid sequence of SEQ ID NO: 2.

2 Claim 26 (Previously Added) The DNA molecule of claim 25 comprising a nucleotide sequence corresponding to SEQ ID NO: 1.

3 Claim 27 (Previously Added) An isolated or recombinant polynucleotide comprising a nucleotide sequence corresponding to SEQ ID NO: 1.

3

4 Claim 28 (Currently Amended) The polynucleotide of claim 9, 27 wherein said polynucleotide is selected from the group comprising:

- (a) RNA;
- (b) cDNA; and
- (c) genomic DNA.

5 Claim 29 (Previously Added) An expression vector comprising one of the DNAs or polynucleotides of claims 25, 26, 27 or 28.

1 2 3 4

5

6 Claim 30 (Previously Added) A cultured cell comprising the expression vector of claim 29.

Remarks

1. Claim 28 has been amended such that it is now dependent from claim 27, not from cancelled claim 9.
2. As detailed in the response dated August 28, 2003 (see in particular amendments to the specifications), the amino acid sequence in Figure 3B is the same as SEQ ID NO: 2 and has been identified as such in the specification. A substitute computer readable form of SEQ ID NO: 2 has already been submitted along with a substitute paper copy of that sequence listing and statement that the content of the two is the same, and that they include no new matter.
3. The Specification and the Figures have been amended so that every reference to Figure 3B has a corresponding reference to the relevant sequence identifier, [SEQ ID NO: 2].